Sanjad Sakati Syndrome

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Sanjad Sakati Syndrome (SSS) is a rare autosomal recessive congenital disorder. It was reported exclusively in people of Arabian origin. SSS comprises of congenital hypoparathyroidism, severe growth retardation, mental retardation and dysmorphic facial features. The typical metabolic derangements lead to several morbid manifestations. SSS is also known as hypoparathyroidism-retardation-dysmorphism (HRD) which was listed in Online Mendelian Inheritance in Mean (OMIM) #241410.

We present the first case of SSS in Bahrain in a 40-days-old female. She was thin and lean. She had a narrow face, deep-seated eyes, peaked nose, long philtrum, thin lips, and micrognathia. She had short stature, small hands and feet, long tapering fingers and clinodactyly.

Parathyroid hormone was 0.3 pmol/L (normal range 0.99-6.05) and and 25-Hydroxy vitamin D was 7 nmol/L (normal range 53-150). The infant is the 4th child of consanguineous parents. The elder male brother who had features suggestive of SSS died at the age of 7 years.

The patient was treated with calcium and vitamin D therapy. Her convulsions were controlled. However, her anthropometric measure did not improve despite aggressive nutritional support via gastrostomy tube feeding. The patient is still alive at the age of 14 years and eight months.

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